



NAGLU gene

N-acetyl-alpha-glucosaminidase

Normal Function

The *NAGLU* gene provides instructions for producing an enzyme called alpha-N-acetylglucosaminidase. This enzyme is located in lysosomes, compartments within cells that digest and recycle different types of molecules. Alpha-N-acetylglucosaminidase is involved in the step-wise breakdown of large molecules called glycosaminoglycans (GAGs). GAGs are composed of sugar molecules that are linked together to form a long string. To break down these large molecules, individual sugars are removed one at a time from one end of the molecule. Alpha-N-acetylglucosaminidase removes a sugar called N-acetylglucosamine when it is at the end of the GAG chain.

Health Conditions Related to Genetic Changes

mucopolysaccharidosis type III

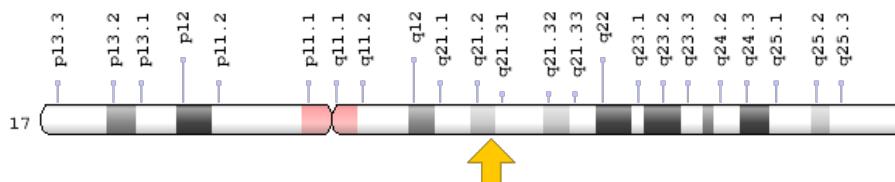
At least 118 mutations in the *NAGLU* gene have been found to cause mucopolysaccharidosis type IIIB (MPS IIIB). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS IIIB reduce or eliminate the function of alpha-N-acetylglucosaminidase.

The lack of alpha-N-acetylglucosaminidase activity disrupts the breakdown of a subset of GAGs called heparan sulfate. As a result, partially broken down heparan sulfate accumulates within lysosomes. Researchers believe that the accumulation of GAGs interferes with the functions of other proteins inside the lysosomes and disrupts the normal functions of cells. It is unknown why the buildup of heparan sulfate mostly affects the central nervous system in MPS IIIB.

Chromosomal Location

Cytogenetic Location: 17q21.2, which is the long (q) arm of chromosome 17 at position 21.2

Molecular Location: base pairs 42,535,933 to 42,544,449 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- alpha-N-acetylglucosaminidase
- alpha-N-acetylglucosaminidase precursor
- ANAG_HUMAN
- N-acetylglucosaminidase, alpha
- N-acetylglucosaminidase, alpha-
- NAG
- UFHSD

Additional Information & Resources

Educational Resources

- Eurekah Bioscience Collection: Defects in Glycosaminoglycan Degradation (Mucopolysaccharidoses)
<https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53462>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NAGLU%5BTIAB%5D%29+OR+%28alpha-N-acetylglucosaminidase%5BTIAB%5D%29+OR+%28N-acetyl-alpha-glucosaminidase%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- N-ACETYLGLUCOSAMINIDASE, ALPHA-
<http://omim.org/entry/609701>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_NAGLU.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NAGLU%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7632
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4669>
- UniProt
<http://www.uniprot.org/uniprot/P54802>

Sources for This Summary

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